

Arginase deficiency

Description

Arginase deficiency is an inherited disorder that causes the amino acid arginine (a building block of proteins) and ammonia to accumulate gradually in the blood. Ammonia, which is formed when proteins are broken down in the body, is toxic if levels become too high. The nervous system is especially sensitive to the effects of excess ammonia.

Arginase deficiency usually becomes evident by about the age of 3. It most often appears as stiffness, especially in the legs, caused by abnormal tensing of the muscles (spasticity). Other symptoms may include slower than normal growth, developmental delays and eventual loss of developmental milestones, intellectual disabilities, seizures, tremors, and difficulty with balance and coordination (ataxia). Occasionally, high-protein meals or stress caused by illness or periods without food (fasting) may cause ammonia to accumulate more quickly in the blood. This rapid increase in ammonia may lead to episodes of irritability, refusal to eat, and vomiting.

In some affected individuals, the signs and symptoms of arginase deficiency may be less severe and may not appear until later in life.

Frequency

Arginase deficiency is a very rare disorder; it has been estimated to occur once in every 300,000 to 1,000,000 individuals.

Causes

Arginase deficiency is caused by variants (also called mutations) in the *ARG1* gene. This gene provides instructions for making an enzyme called arginase.

Arginase deficiency belongs to a class of genetic diseases called urea cycle disorders. The urea cycle is a sequence of reactions that occurs in liver cells. This cycle processes excess nitrogen, generated when protein is used by the body, to make a compound called urea that is excreted by the kidneys.

The arginase enzyme facilitates the final step of the urea cycle, which produces urea by removing nitrogen from arginine. In people with arginase deficiency, arginase is damaged or missing, and arginine is not broken down properly. As a result, urea cannot be produced normally, and excess nitrogen accumulates in the blood in the form of

ammonia. The accumulation of ammonia and arginine are believed to cause the neurological problems and other signs and symptoms of arginase deficiency.

[Learn more about the gene associated with Arginase deficiency](#)

- ARG1

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell must have a variant to cause the disorder. The parents of an individual with an autosomal recessive condition each carry one copy of the altered gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- ARG1 deficiency
- Arginase deficiency disease
- Argininemia
- Hyperargininemia

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Arginase deficiency (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0268548/>)

Genetic and Rare Diseases Information Center

- Argininemia (<https://rarediseases.info.nih.gov/diseases/5840/index>)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

Clinical Trials

- ClinicalTrials.gov ([https://clinicaltrials.gov/search?cond=%22Arginase deficiency%22](https://clinicaltrials.gov/search?cond=%22Arginase+deficiency%22))

Catalog of Genes and Diseases from OMIM

- ARGININEMIA (<https://omim.org/entry/207800>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28arginase+deficiency%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>)

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